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CASE REPORT

A CASE REPORT OF APHALLIA

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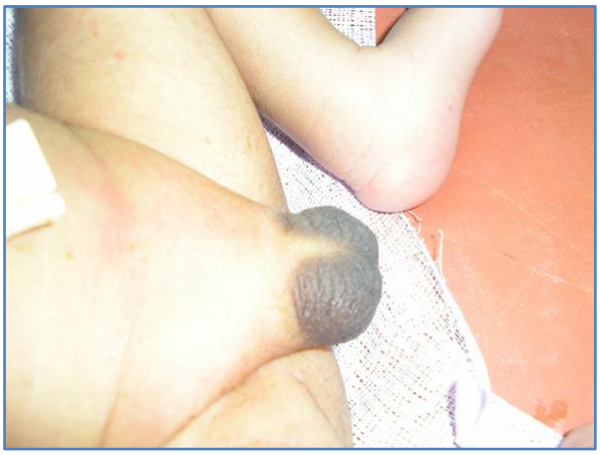
ABSTRACT: Aphallia is a complex urogenital malformation with an occurrence of 1 in every 30 million births; only 75 cases have been reported in the literature till recently [1, 2]. A one month old infant was brought to our hospital with aphallia. The child was born of an uncomplicated pregnancy with no family history of any other congenital anomalies or consanguinity. Examination revealed a well-child with an absent penis. The child had a normal scrotum with bilaterally descended testis. There was absence of urethral meatus in the perineum.

KEY WORDS: Aphallia, phalloplasty, genital tubercle

We are hereby reporting a case of Aphallia which affected an infant whose parents have decided to keep the male sex.

CASE REPORT: HISTORY: A one month old infant brought to our hospital for absence of penis. The pregnancy that led to his birth was at term of an uncomplicated pregnancy with no family history of any other congenital anomalies or consanguinity.The child's mother was a young normal lady with no history of use of any drugs or exposure to x-ray during pregnancy.



Arrow Shows absence of penis with scrotum

CLINICAL EXAMINATION: showed a good general state, an absence of penis and urinary meatus.[3] Palpation revealed the presence of the gonads inside the scrotum. The anus was in normal position, permeable, with the presence of feces mixed with urine.

INVESTIGATIONS: Standard abdomen X-ray and abdomen CT scan were normal. Complete blood count, liver and renal function tests were normal.

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The neonate was masculine, 46 karyotype, XY.[4] Laboratory tests including hormonal assay were unremarkable. Barium meal and enema study revealed normal gastro-intestinal tract pattern. Ultrasonography of abdomen and IVP reveals normal kidneys.

It was planned that the child would undergo phalloplasty at the age of puberty because his parents decided to keep the male sex.

DISCUSSION: Aphallia is a congenital malformity in which the phallus (penis or clitoris) is absent. The word is derived from the Greek "a" for negative or no, and "phallia" for penis. It is an anomaly which occurs during the fourth week of embryonic development, which accounts for its frequent association with related malformations. It is related to result from the non-formation of the genital tubercle or its failure to develop. A genital tubercle or phallic tubercle is a body of tissue present in the development of the urinary and reproductive organs.

It forms in the ventral, caudal region of mammalian embryos of both sexes, and eventually develops into a phallus. In the human fetus, the genital tubercle develops around week 4 of gestation, and by week 9 becomes recognizably either a clitoris or penis. This should not be confused with the sinus tubercle which is a proliferation of endoderm induced by paramesonephric ducts.[5] Even after the phallus is developed, the term genital tubercle remains, but only as the terminal end of it, which develops into either the glans penis or the glans clitoridis.

The genital tubercle is sensitive to dihydrotestosterone and rich in 5-alpha-reductase, so that the amount of fetal testosterone present after the second month is a major determinant of phallus size at birth.[6] Its diagnostic is easy at birth, as there is an absence of penis. However, some cases were diagnosed late.

Treatment generally performed is phalloplasty or feminizing genitoplasty.[3]

The birth of a child affected by aphallia puts parents in a highly delicate situation. It is the responsibility of the medical doctor to reassure them by proposing therapeutic options, that is, feminizing genitoplasty or phalloplasty. No surgical intervention should be undertaken before counselling the parents.

However, despite of the practical problems of reconstructing a normally functioning penis, one of the options is rearing such children as males because the social stigma for females in our society may be much greater. Unmarried male can be far better in life than an unmarried female. So, congenital aphallia is an extremely rare condition that requires multidisciplinary and individualized management.

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